

# PAX3

## Mouse Mutants

### MGI database of Pax3 mutants

*Pax3<sup>Sp</sup>*: Spontaneous; original classic Splotch mutation; mutation in intron 3 resulting in 4 different, aberrantly spliced and truncated mRNAs (Russell and Roscoe, 1947)

*Pax3<sup>Sp-1H</sup>*: Radiation induced (Beechey and Searle, 1986)

*Pax3<sup>Sp-2H</sup>*: Radiation induced; 32 bp deletion in the paired homeodomain (Beechey and Searle, 1986, Epstein et al., 1991b)

*Pax3<sup>Sp-r</sup>*: Radiation induced; homozygotes die before implantation; heterozygotes show growth retardation through adulthood; harbor a 14-16 cM deletion of *Pax3* and surrounding loci (Beechey and Searle, 1986, Epstein et al., 1991a)

*Pax3<sup>Sp-J</sup>*: Spontaneous (Dickie, 1964); similar phenotype to Sp; extinct

*Pax3<sup>Sp-2J</sup>*: Spontaneous (Dickie, 1964); similar phenotype to Sp; extinct

*Pax3<sup>Sp-3J</sup>*: Spontaneous (Dickie, 1964); similar phenotype to Sp; extinct

*Pax3<sup>Sp-4H</sup>*: Radiation induced deletion (Goulding et al., 1993)

*Pax3<sup>Sp-5H</sup>*: Radiation induced, semidominant (Rasberry and Cattanach, 1994)

*Pax3<sup>Sp-6H</sup>*: Chemically and Radiation induced, semidominant (Cattanach et al., 1994)

*Pax3<sup>Sp-7H</sup>*: ENU induced; point mutation resulted in V38G substitution, which is the first amino acid of the paired domain (Bogani et al., 2004)

*Pax3<sup>Sp-d</sup>*: Spontaneous; paired domain point mutation; Named Splotch-delayed, because death of homozygotes is delayed to late in gestation; display caudal but not cranial rachischisis (exposure of neural tube) (Dickie, 1964)

*Pax3<sup>tm1(cre)Joe</sup>*: Targeted knock-in (Lang et al., 2005)

*Pax3<sup>tm1(FOXO1A)Gcg</sup>*: Targeted knock-in of a fragment of the PAX3-FKHR fusion protein, to model alveolar rhabdomyosarcoma (Lagutina et al., 2002)

*Pax3<sup>tm1.1Sjc</sup>*: Targeted deletion of exon containing two-thirds of the homeodomain (Koushik et al., 2002)

*Pax3<sup>tm1.2Sjc</sup>*: Targeted deletion of exon containing two-thirds of the homeodomain (Koushik et al., 2002)

*Pax3<sup>tm1Buck</sup>*: Targeted; LacZ reporter gene replacement (Relaix et al., 2003)

*Pax3<sup>tm1Mrc</sup>*: Conditional targeted knock-in of Foxo1 gene at 3' end of *Pax3*, to model alveolar rhabdomyosarcoma (Keller et al., 2004)

*Pax3<sup>tm1Sjc</sup>*: Targeted (floxed/Frt); allows conditional knockout of homeodomain (Koushik et al., 2002)

*Pax3<sup>tm2</sup>(PAX3/FOXO1A)<sup>Buck</sup>*: Targeted (knock-in); puromycin, human *PAX3-FOXO1A* and *LacZ* cassette insertion ([Relaix et al., 2003](#))

*Pax3<sup>tm2.1</sup>(PAX3/FOXO1A)<sup>Buck</sup>*: Targeted (knock-in); human *PAX3-FOXO1A* and *LacZ* cassette insertion at *Pax3* locus ([Relaix et al., 2003](#))

*Pax3<sup>tm3</sup>(Pax7)<sup>Buck</sup>*: Targeted (knock-in); insertion of *Pax7* at *Pax3* locus; full construct prior to Cre excision of blocking upstream sequences ([Relaix et al., 2004](#))

*Pax3<sup>tm3.1</sup>(Pax7)<sup>Buck</sup>*: Targeted (knock-in); insertion of *Pax7* at *Pax3* locus; *Pax3* expression still detected in this line ([Relaix et al., 2004](#))

*Pax3<sup>tm4</sup><sup>Buck</sup>*: Targeted (reporter); insertion of EGFP at exon 1 ([Relaix et al., 2005](#))

*Pax3<sup>Cre</sup>*: Targeted insertion of Cre at exon 1 of *Pax3* ([Engleka et al., 2005](#))

*Pax3<sup>tm5.1</sup><sup>Buck</sup>*: Targeted (reporter); insertion of construct encoding a GFP-FRT-Puro cassette followed by a fusion protein consisting of the PAX3 DNA-binding domain and the engrailed repressor domain; Cre recombination allows fusion protein expression ([Bajard et al., 2006](#))

*Pax3<sup>tm5</sup><sup>Buck</sup>*: Targeted (Floxed/Frt); insertion of a construct encoding a fusion protein consisting of the PAX3 DNA-binding domain and the engrailed repressor domain ([Bajard et al., 2006](#))

*Pax3<sup>wbs</sup>*: ENU induced; nonsense mutation results in truncation at paired domain, no protein formed, so functional null ([Guo et al., 2010](#))

*Pax3<sup>tm2</sup><sup>Joe</sup>*: Targeted deletion of upstream enhancer region, with retention of floxed PGK-neo cassette. This cassette interferes with *Pax3* transcription, and can be removed for tissue specific rescue ([Degenhardt et al., 2010](#))

*Pax3<sup>tm2.1</sup><sup>Joe</sup>*: CRE recombination-mediated removal of upstream enhancer floxed PGK-neo cassette in *Pax3<sup>tm2</sup><sup>Joe</sup>* mutant; mice show no phenotypic differences from normal mice ([Degenhardt et al., 2010](#))

*Pax3<sup>mi1</sup><sup>Nisw</sup>*: ENU induced; A312T missense mutation (MGI direct submission)

## Transgenics:

*Tg(Pax3-cre)1<sup>Joe</sup>*: Transgene insertion ([Li et al., 2000](#))

*Tg(Pax3-cre)2<sup>Joe</sup>*: Transgene insertion ([Li et al., 2000](#))

*Gt(ROSA)26Sor<sup>tm2</sup>(Pax3)<sup>Joe</sup>*: Transgene containing floxed neo and stop codon followed by *Pax3* cDNA; CRE-mediated removal of stop allowed tissue-specific expression ([Wu et al., 2008](#))